

Product datasheet for **SC128237**

SMN1 (NM_000344) Human Untagged Clone

Product data:

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|---------------------------|---|
| Product Type: | Expression Plasmids |
| Product Name: | SMN1 (NM_000344) Human Untagged Clone |
| Tag: | Tag Free |
| Symbol: | SMN1 |
| Synonyms: | BCD541; GEMIN1; SMA; SMA1; SMA2; SMA3; SMA4; SMA@; SMN; SMNT; T-BCD541; TDRD16A |
| Mammalian Cell Selection: | None |
| Vector: | <u>pCMV6-XL5</u> |
| E. coli Selection: | Ampicillin (100 ug/mL) |

Fully Sequenced ORF: >OriGene sequence for NM_000344 edited
GGCCCCACGCTGCGCACCCGCGGTTTGCTATGGCGATGAGCAGCGGCGGCAGTGGTGGC
GGCGTCCCGGAGCAGGAGGATTCCGTGCTGTTCCGGCGCGGCACAGGCCAGAGCGATGAT
TCTGACATTTGGGATGATACAGCACTGATAAAAGCATATGATAAAGCTGTGGCTTCAATTT
AAGCATGCTCTAAAGAATGGTGACATTTGTGAAACTTCGGGTAAACCAAAAACACACCT
AAAAGAAAACCTGCTAAGAAGAATAAAAGCAAAAGAAGAATACTGCAGCTTCTTACAA
CAGTGGAAAGTTGGGACAAATGTTCTGCCATTTGGTCAGAAGACGGTTGCATTTACCCA
GCTACCATTGCTCAATTGATTTAAGAGAGAAACCTGTGTTGTGGTTTACTGGATAT
GGAAATAGAGAGGAGCAAAATCTGTCCGATCTACTTCCCAATCTGTGAAGTAGCTAAT
AATATAGAACAGAATGCTCAAGAGAATGAAATGAAAGCCAAGTTTCAACAGATGAAAGT
GAGAACTCCAGGTCTCCTGGAAATAAATCAGATAACATCAAGCCAAATCTGCTCCATGG
AACTCTTTTCTCCCTCCACCACCCCATGCCAGGGCCAAGACTGGGACCAGGAAAGCCA
GGTCTAAAATTAATGGCCACCACCGCCACCACCACCACCACCCCACTTACTATCA
TGCTGGTGCCTCCATTTCTTCTGGACCACCAATAATCCCCACCACCTCCCATATGT
CCAGATTCTCTTGATGATGCTGATGCTTTGGGAAGTATGTTAATTTATGGTACATGAGT
GGCTATCATACTGGCTATTATATGGGTTTTAGACAAAATCAAAAAGAAGGAAGGTGCTCA
CATTCTTAAATTAAGGAGAAATGCTGGCATAGAGCAGCACTAAATGACACCACTAAAGA
AACGATCAGACAGATCTGGAATGTGAAGCGTTATAGAAGATAACTGGCCTCATTTCTTCA
AAATATCAAGTGTGGGAAAGAAAAAGGAAGTGAATGGGTAACCTTTCTTATGATTA
GTTATGTAATAACCAAAATGCAATGTGAAATATTTTACTGGACTCTATTTTGAAAAACCAT
CTGTAAGAACTGAGGTGGGGTGGGAGGCCAGCAGGTGGTGAAGCAGTTGAGAAAAAT
TGAATGTGGATTAGATTTTGAATGATATTGGATAATTATTGGTAATTTTATGAGCTGTGA
GAAGGGTGTGTAGTTTATAAAGACTGTCTAATTTGCATACTTAAGCATTTAGGAATG
AAGTGTAGAGTGTCTTAAATGTTTCAAATGGTTTAAACAAAATGTATGTGAGGCGTATG
TGGCAAAATGTTACAGAATCTAACTGGTGGACATGGCTGTTTCTTGTACTGTTTTTTCT
ATCTTCTATATGTTTAAAGTATATAATAAAAAATTTTAAATTTTTTTTTTAAAAA
AAAAAAAAAAAAAAAAAAAAAAAAA



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|-------------------------------------|---|
| 5' Read Nucleotide Sequence: | >OriGene 5' read for NM_000344 unedited GTTTTCATTTGTATACGACTCATATAGGCGGCCGCGNATTCAGATCTGGTACCGGTCCGG AATTCCCGGGATATCGTCGACCCACGCGTCCGGGCCACGCTGCCACCCGCGGGTTTG CTATGGCGATGAGCAGCGCGGCAGTGGTGGCGGCGTCCCGGAGCAGGAGGATCCGTGC TGTTCCGGCGCGGCACAGGCCAGAGCGATGATTCTGACATTTGGGATGATACAGCACTGA TAAAAGCATATGATAAAGCTGTGGCTTCATTTAAGCATGCTCTAAAGAATGGTGACATTT GTGAACTTCGGGTAAACCAAAAACACACCTAAAAGAAAACCTGCTAAGAAGAATAAAA GCCAAAAGAAGAATACTGCAGCTTCCTTACAACAGTGGAAGTTGGGGACAAATGTTCTG CCATTTGGTCAGAAGACGGTTGCATTTACCCAGCTACCATTGCTTCAATTGATTTAAGA GAGAACTGTGTTGTGGTTTACTACTGGATATGGAAATAGAGAGGAGCAAAATCTGTCCG ATCTACTTTCCCAATCTGTGAAGTAGCTAATAATATAGAACAGAATGCTCAAGAGAATG AAAATGAAAGCCAAGTTTCAACAGATGGAAAGTGAGAACTCCAGGTCTCTGAAAAAT AAATCCAGATTAACATCAAGGCCAAAACTTGCTCCATGGGAACTCTTTTTTTTCCCGC CACCACCCCATGCCAAGGGCCAAGACTGGGGACCCAGGAAAGCCAGGTTTAATATTTA ATGGGCCACCACCGGTCTCCGTACCCACCACCCCCCATTTACCATTAAGCCTGGC TGGCCTCCATTTCTTTGGGGACCCCCACATAATTTTC |
| Restriction Sites: | Please inquire |
| ACCN: | NM_000344 |
| Insert Size: | 1500 bp |
| OTI Disclaimer: | Our molecular clone sequence data has been matched to the reference identifier above as a point of reference. Note that the complete sequence of our molecular clones may differ from the sequence published for this corresponding reference, e.g., by representing an alternative RNA splicing form or single nucleotide polymorphism (SNP). |
| Components: | The ORF clone is ion-exchange column purified and shipped in a 2D barcoded Matrix tube containing 10ug of transfection-ready, dried plasmid DNA (reconstitute with 100 ul of water). |
| Reconstitution Method: | <ol style="list-style-type: none"> 1. Centrifuge at 5,000xg for 5min. 2. Carefully open the tube and add 100ul of sterile water to dissolve the DNA. 3. Close the tube and incubate for 10 minutes at room temperature. 4. Briefly vortex the tube and then do a quick spin (less than 5000xg) to concentrate the liquid at the bottom. 5. Store the suspended plasmid at -20°C. The DNA is stable for at least one year from date of shipping when stored at -20°C. |
| RefSeq: | NM_000344.2 , NP_000335.1 |
| RefSeq Size: | 1621 bp |
| RefSeq ORF: | 885 bp |
| Locus ID: | 6606 |
| UniProt ID: | Q16637 |
| Cytogenetics: | 5q13.2 |
| Protein Families: | Druggable Genome, Stem cell - Pluripotency |

Gene Summary:

This gene is part of a 500 kb inverted duplication on chromosome 5q13. This duplicated region contains at least four genes and repetitive elements which make it prone to rearrangements and deletions. The repetitiveness and complexity of the sequence have also caused difficulty in determining the organization of this genomic region. The telomeric and centromeric copies of this gene are nearly identical and encode the same protein. However, mutations in this gene, the telomeric copy, are associated with spinal muscular atrophy; mutations in the centromeric copy do not lead to disease. The centromeric copy may be a modifier of disease caused by mutation in the telomeric copy. The critical sequence difference between the two genes is a single nucleotide in exon 7, which is thought to be an exon splice enhancer. Note that the nine exons of both the telomeric and centromeric copies are designated historically as exon 1, 2a, 2b, and 3-8. It is thought that gene conversion events may involve the two genes, leading to varying copy numbers of each gene. The protein encoded by this gene localizes to both the cytoplasm and the nucleus. Within the nucleus, the protein localizes to subnuclear bodies called gems which are found near coiled bodies containing high concentrations of small ribonucleoproteins (snRNPs). This protein forms heteromeric complexes with proteins such as SIP1 and GEMIN4, and also interacts with several proteins known to be involved in the biogenesis of snRNPs, such as hnRNP U protein and the small nucleolar RNA binding protein. Multiple transcript variants encoding distinct isoforms have been described. [provided by RefSeq, Jul 2014]

Transcript Variant: This variant (d) represents the longest transcript and encodes the longest isoform (d). This variant is thought to be the predominant transcript produced by this copy of the gene.